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Unusual recovery of respiratory chain complex-III deficiency upon G-tube feeding and a cocktail

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Letter to the Editor

With interest we read the article by Mori et al. about a 7 year old male with respiratory-chain complex-III-deficiency due to a 9 bp-deletion in the cytb gene, which initially manifested with failure to thrive, vomiting, diarrhea, constipation, weight-loss, fatigue, exercise-intolerance, muscle weakness, metabolic acidosis, and creatine-kinase elevation and who recovered almost completely by age 12 years [1]. We have the following comments and concerns.

In the majority of the cases mitochondrial disorders (MIDs) are progressive [2]. Thus, the marked clinical improvement is quite unusual. Unfortunately, the results of the clinical neurologic exam were not provided but obviously the patient initially had muscle weakness since he was wheelchair-bound [1]. How to explain complete recovery of weakness particularly with a heteroplasmy rate > 90% in the muscle? Why do the authors attribute recovery to G-tube-feeding and not to the cocktail of coenzyme-Q, L-carnitine, cornstarch, riboflavin, creatine-monohydrate, alpha-lipoic-acid, and medium-chain triglycerides? Is it conceivable that there was secondary coenzyme-Q-deficiency or carnitine-deficiency? Did consolidation of the phenotype persist even after discontinuation of G-tube-feeding and the cocktail? Was the patient seen for follow-up after finishing treatment? Were coenzyme-Q levels reduced before substitution?

Nothing is reported about the clinical presentation of the mother and the patient's siblings [1]. Was the cytb deletion also found in the mother or any sibling? Were clinical manifestations of the index case variable from those of his siblings or his mother?

Did the patient ever experience a seizure or a stroke-like-episode? Was an EEG recorded? Were cognitive functions impaired? Did he have a history of migraine? Was there steatosis, cysts, or hepatomegaly?

Overall, this interesting case merits a broader discussion about the possible mechanisms of recovery and a more profound description of the phenotype and disease course. Furthermore, clinical and genetic data about his relatives are essential as well as follow-up data after discontinuation of G-tube-feeding and the cocktail.

References

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